

## CASE STUDY

### CONSTRUCTING A PEDIGREE CHART

#### Objective

To construct and examine a pedigree chart for Duchenne's muscular dystrophy, a sex-linked condition in humans.

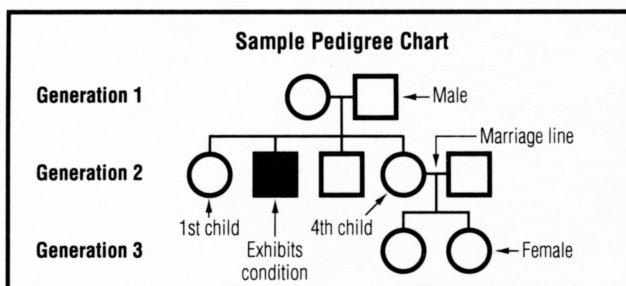
#### Background Information

Duchenne's muscular dystrophy is a well-known lethal condition. Evidence supports its inclusion as a sex-linked condition, as it appears nearly always in males. It is usually carried by females. The disease causes the muscles of the body to slowly deteriorate (atrophy). Boys with this condition often need to use a wheelchair by five years of age, and death usually occurs in the early to mid-teens. Knowing how this disease is inherited through family members suspected of carrying the gene will be helpful in genetic counselling and decision making.

#### Procedure

**Note:** The symbol DMD will be used to represent Duchenne's muscular dystrophy.

- 1 A pedigree chart (see *Nelson Biology*, pages 595, 616, and 660) attempts to trace a specific trait using a series of circles, squares, and straight lines to represent generations in a family. Circles represent females, squares represent males, and lines join individuals. Shaded circles and squares indicate individuals who show the condition being studied. Children of a union are listed in order from left to right and generations are listed chronologically from top to bottom.



- 2 Carefully read the following information concerning four generations of a family.

- Two people marry and have four children ( $F_1$  generation) in the following order: daughter, son, daughter, daughter.
- The **first daughter** ( $F_1$ ) marries and has three children: a son, a daughter, a second daughter. Her son does not marry. Her first daughter does not marry. Her second daughter marries and produces a son, who develops DMD, and two daughters.

*This family history is designated Line A.*

- The **son** ( $F_1$ ) develops muscular dystrophy and dies.
- The **second daughter** ( $F_1$ ) marries, but produces no children.
- The **third daughter** ( $F_1$ ) marries and has two children: a son who develops DMD and dies, and a second son. Her second son marries. His wife produces two children: a daughter and then a son.

*This family history is designated Line B.*

- a) Construct a pedigree chart to represent the information provided. The chart should occupy at least half a page. Keep Line A and Line B on opposite sides of the page. Mark in the generations.
- 4 Check the accuracy of your pedigree chart with your teacher before proceeding with the questions below.
    - b) If the original parents did not exhibit the trait for DMD, how could it have appeared in one of their children?
    - c) Is there any indication from the pedigree chart that DMD is a sex-linked condition? Explain.
    - d) Which females in generation 2 are definitely carriers of the DMD condition? Explain.
    - e) In Line A, generation 3, could the husband of the married daughter have been responsible for passing the DMD gene to his son? Explain.

#### Application Questions

- 1 Is it possible for a sex-linked disease like DMD to pass from one generation to another without appearing in any offspring? Explain.
- 2 In Line B, generation 4, what is the probability of the son developing DMD? Explain your answer.
- 3 In generation 2, concerned by the appearance of DMD in her brother, the second daughter and her husband decided to visit a genetic counsellor before having any children. Their decision was to adopt rather than have children of their own. Reconstruct the reasoning they probably used in arriving at this decision. ■